Recessive TBC1D24 Mutations Are Frequent in Moroccan Non-Syndromic Hearing Loss Pedigrees

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Résumé en anglais	Mutations in the TBC1D24 gene are responsible for four neurological presentations: infantile epileptic encephalopathy, infantile myoclonic epilepsy, DOORS (deafness, onychodystrophy, osteodystrophy, mental retardation and seizures) and NSHL (non-syndromic hearing loss). For the latter, two recessive (DFNB86) and one dominant (DFNA65) mutations have so far been identified in consanguineous Pakistani and European/Chinese families, respectively. Here we report the results of a genetic study performed on a large Moroccan cohort of deaf patients that identified three families with compound heterozygote mutations in TBC1D24. Four novel mutations were identified, among which, one c.641G>A (p.Arg214His) was present in the three families, and has a frequency of 2% in control Moroccan population with normal hearing, suggesting that it acts as an hypomorphic variant leading to restricted deafness when combined with another recessive severe mutation. Altogether, our results show that mutations in TBC1D24 gene are a frequent cause (>2%) of NSHL in Morocco, and that due to its possible compound heterozygote recessive transmission, this gene should be further considered and screened in other deaf cohorts.

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